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Molecular Cytogenetics and Gene Analysis: Implications for Oncology Nurses

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Recent advances in genetics have provided a better understanding of many diseases, including cancer. These advances will have an impact on oncology clinical practice in an unprecedented way because they will improve cancer detection and diagnosis. Molecular cytogenetic techniques, such as fluorescent in situ hybridization (FISH), comparative genomic hybridization (CGH), and spectral karyotyping (SKY), analyze chromosomes and their abnormalities at the molecular level. The application of these techniques to disease diagnostics also will impact treatment modalities and prognosis.

Nurses must be knowledgeable about the rapid expansion of genetic information as it relates to the diagnosis and treatment of cancer. Oncology nurses are in a unique position to translate this information to patients and their families.

Gene Structure and Function

Deoxyribonucleic acid (DNA) is a double-stranded molecule found within the nucleus of each cell. DNA contains four complementary base pairs: adenine, thymine, guanine, and cytosine. Like letters are arranged to form words, the sequence of the base pairs is arranged to encode genes. DNA is the genetic "blueprint" for all proteins in the body, providing the instructions for cell growth,

Current advances in genetics have provided a better understanding of many diseases, including cancer, and will have an impact on oncology clinical practice in an unprecedented way. The molecular cytogenetic techniques of fluorescent in situ hybridization (FISH), comparative genomic hybridization (CGH), and spectral karyotyping (SKY) are providing tremendous insights into genetic information related to cancer by specifically illustrating chromosomal abnormalities that can occur in a patient's cancer cells. The application of these techniques allows for the development of molecular diagnostic tumor-specific markers. These molecular diagnostic tests may be applied to clinical material, which may help to improve the diagnosis and staging of a patient's tumor, particularly in small, premalignant lesions that often are equivocal and difficult to assess. An understanding of these genetic changes will provide a foundation of knowledge for oncology nurses that will lead to significantly improved detection methods, therapies, and disease prevention. As members of the healthcare team, oncology nurses must be knowledgeable about the rapid expansion of genetic information. Oncology nurses are in a unique position to translate this information to patients and their families and, ultimately, enhance comprehensive care through patient education and advocacy.

differentiation, function, and death (Jorde, Carey, Bamshad, & White, 1999).

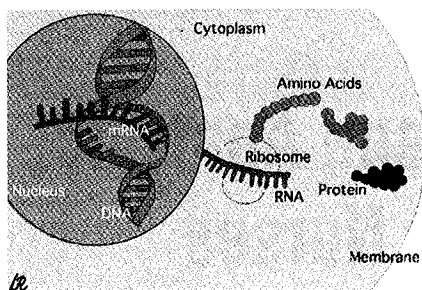
The genetic information needed for the basic functions of the cell is converted into proteins by a process called protein synthesis. Protein synthesis is characterized by two essential steps—transcription and translation. During transcription, a fragment of the double-stranded DNA separates and is cop-

ied into messenger ribonucleic acid (mRNA). The mRNA assembles itself along the single-stranded DNA by attaching its complementary base pairs to the DNA. The sequence of the mRNA is determined by the order of base pairs along the DNA. Following this step, the mRNA contains the information necessary to construct the amino acid sequence to make a protein. The mRNA leaves the DNA and carries this information out of the nucleus to a ribosome in the cytoplasm. At this point, the information is ready for translation, where it is decoded and assembled into amino acid chains, forming proteins (see Figure 1) (Jorde et al., 1999).

Transcription and translation are regulated by hormones, proteins, or DNA sequences called enhancers, which may be located upstream or downstream on the gene (Jorde et al., 1999; Lea, Jenkins, & Francomano, 1998). Whereas enhancers function to increase transcription of specific genes within the cell nucleus, other DNA sequences known as si-

lencers function to suppress the transcription of genes. Almost all cells contain the exact same DNA; however, certain genes are transcribed in specific tissues at various times. This explains why a large variability exists among cells even though they contain

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During the first step (transcription), the DNA is separated and copied into messenger RNA (mRNA). The second step (translation) takes place at a ribosome in the cytoplasm after the mRNA leaves the nucleus. At the ribosome, the mRNA is decoded and assembled into amino acid chains, forming proteins.

FIGURE 1. PROTEIN SYNTHESIS

the same DNA (Jorde et al.). The variability in gene expression and protein synthesis is what influences cell growth and differentiation (Lea et al.).

Chromosomes and Cancer

If DNA from a single cell was uncoiled, it would measure two meters in length. However, DNA is packaged tightly and held together by histone proteins forming structures called chromosomes (Jorde et al., 1999). Chromosomes are like volumes of books because they are the organizing units of genetic material in a cell. During cell division, chromosomes replicate and transfer the genetic information to two daughter cells. The morphology, or shape, of chromosomes changes during the cell cycle. During most stages of the cell cycle, chromosomes are elongated and difficult to differentiate; however, they can be visualized best during mitosis, when the cell is preparing for cell division and the chromosomes condense. Figure 2 shows an example of condensed metaphase chromosomes.

Diploid human somatic cells contain 46 chromosomes, which are 22 pairs of autosomes, or non-sex chromosomes, and two sex chromosomes, X and Y. The fact that diseases such as cancer can be correlated to particular changes in the chromosomes has created interest in chromosome analysis for diagnostic purposes. Chromosomes can be visualized through a microscope and are distinguishable according to size, shape, and their banding pattern. These specific characteristics allow for identification of chromosomal aberrations. Unstable chromosomes can cause cancer, and cancer progression often is characterized by an increasing number of acquired chromosomal aberrations.

Multiple steps are required for the full development of cancer (Therman & Susman, 1993). The steps include changes in chromosome number and structure and gene mutations. Changes in chromosomal number, or aneuploidy, occur when entire chromosomes are gained or lost (Lea et al., 1998). Aneuploidy is seen in most tumors, and the distribution of such changes is specific for different tumors. For example, in breast cancer, a trisomy, or extra copy, of chromosomes 7, 8, 18, and 20 often exists (Heim & Mitelman, 1995).

Changes in chromosomal structure include translocations, deletions, and inversions (Jorde et al., 1999). A translocation is the exchange of genetic material from one chromosome to another. This can result in the activation of a gene that causes the cell to progress to cancer. In the case of chronic myelogenous leukemia, a translocation occurs in most cases between chromosomes 9 and 22 (Rowley, 1990), resulting in the generation of a fusion protein that causes malignant transformation.

A deletion is a loss of DNA from a chromosome. For instance, a deletion of a particular DNA segment on chromosome 13 is the cause of retinoblastoma, an eye tumor (Goddard et al., 1988; Weinberg, 1992). An inversion is a segment of DNA reversed within a chromosome and is found in acute myelogenous leukemias among many other cancers (Heim & Mitelman, 1995).

Chromosomal analysis, therefore, allows one to visualize genetic aberrations on the chromosomal level. It is important, however, to understand that the consequences of chromosomal aberrations are defects in specific genes. These genes include oncogenes and tumor-suppressor genes.

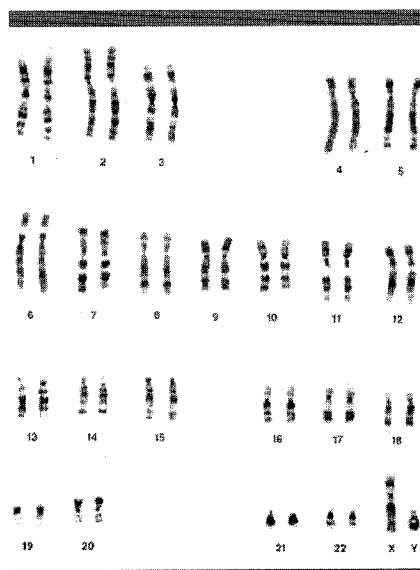


FIGURE 2. A BANDED KARYOTYPE ILLUSTRATING CONDENSED METAPHASE CHROMOSOMES OF A NORMAL MALE (46, XY)

Oncogenes

Within each normal cell are genes called oncogenes, whose normal activity promotes cell proliferation (Strachan & Read, 1999). Oncogenes are turned on and off during embryonic development at different steps of the cell cycle and in different cell types (DeVita, Hellman, & Rosenberg, 2001). When mutated, these genes become overactive. Abnormal, unrestrained stimulation of cell proliferation occurs, which can help to transform a normal cell into a tumor cell (DeVita et al.; Strachan & Read). The nonmutant forms are properly called "proto-oncogenes"; however, the term "oncogene" now is widely used for both the normal and activated forms of such genes (Strachan & Read). To use an analogy, oncogenes function like the gas pedal of a car. When mutated, the gas pedal jams and the car drives out of control.

Activation of oncogenes can occur by point mutations, chromosomal translocations, or DNA amplification. A point mutation is a common DNA alteration in cancer. It is a change in a single-base pair that can alter one amino acid in a protein. If a point mutation occurs in a critical region of a gene, the faulty protein can result in a disturbance in the regulation of cell division (Varmus & Weinberg, 1993). In the event of a chromosomal translocation, an inactive oncogene may become active because it has been moved to the vicinity of an activating gene (Varmus & Weinberg). Finally, gene amplification can alter cell proliferation, which occurs when a single gene is reduplicated into hundreds of copies. Normally, cells contain two copies of a gene; however, in gene amplification, multiple copies of the gene are found in a single region along a chromosome, dispersed on other chromosomes, or, in some cases, forming their own minute chromosomes (Varmus & Weinberg). This amplified DNA increases the amount of RNA and protein within a cell, and the stimulated growth activity results in uncontrolled cell proliferation, leading to tumor development (Varmus & Weinberg). A well-established example of an amplified oncogene involved in breast cancer is HER2 (*c-erbB-2*). This oncogene is overexpressed in 20%–30% of breast cancers and is associated with a poor clinical outcome (Harris et al., 2001). The detection of an amplification of this oncogene by FISH also stratifies patients into groups that receive a specific treatment.

Tumor-Suppressor Genes

Tumor-suppressor genes normally function to suppress or control cell division (Strachan & Read, 1999). Their gene prod-

Glossary

Allele: A gene or particular DNA sequence at a specific chromosomal location. Each individual possesses two alleles of each gene or specific DNA sequence—one inherited from the father and one from the mother.

Aneuploidy: A chromosome constitution with one or more chromosomes extra or missing from a full set of 46 chromosomes in humans.

Anneal: The association of complementary DNA strands to form a double helix.

Autosome: Any chromosome other than the sex chromosomes, X and Y.

Denature (denaturation): Dissociation of complementary strands of the double helix to create single-stranded DNA.

Diploid: Having two copies of each type of chromosome (46 total in human); the normal constitution of most human somatic cells.

Dominant: Describes any trait that is expressed heterozygously (Aa).

Fusion protein: The product of a gene containing a coding sequence from two different genes, often resulting from a chromosomal translocation.

Genome: The total genetic complement of an organism or virus.

Haploid: Describing a cell (typically a sex cell) that has only a single copy of each chromosome (23 in humans).

Heterozygous: Explains if an individual has two different alleles for a particular gene or trait, written as Aa.

Homologous chromosomes: The two copies of a chromosome in a diploid cell. Unlike sister chromatids, homologous chromosomes are not copies of each other; one was inherited from the father and the other from the mother.

Homozygous: An individual is homozygous for a particular gene if he or she has two identical alleles for that gene. For example, a person often is described as homozygous AA or aa if he or she has two normally functioning or two pathogenic alleles.

Hybridize (hybridization): Testing for the presence of a given sequence in a DNA sample by mixing single DNA strands from a known probe with single DNA strands of the target sample then allowing complementary strands to anneal.

Karyotype: A summary of the chromosome constitution of a cell or person, such as 46, XY.

Morphology: Describes the shape of a cell and its counterparts.

Metaphase chromosomes: During the metaphase stage of cell division, the chromosomes condense and line up on the equatorial plane of the cell, preparing to be divided equally into two daughter cells.

Oncogene: A gene involved in control of cell proliferation, which, when overactive, can help to transform a normal cell into a tumor cell. Originally, the word was used only for the activated forms of the gene, and the normal cellular gene was called a proto-oncogene. This distinction now is widely ignored.

Point mutation: A change in a single-base pair of a gene that consequently can alter the protein expressed by the gene.

Probe: A known DNA fragment of interest labeled with fluorescence, which is used in hybridization experiments to identify complementary DNA sequences.

Proto-oncogene: A cellular gene, which can be converted by activating mutations into an oncogene. The term oncogene now is widely used for both the normal and activated forms of such genes.

Recessive: Describes any trait that is expressed homozygously (aa).

Tumor-suppressor (TS) genes: Genes whose normal function is to inhibit or control cell division. Tumors always have inactivating mutations in TS genes.

Note. Based on information from Strachan & Read, 1999.

a consequence of only one defective, dominant gene.

Mutated tumor-suppressor genes are involved in the majority of tumors (Strachan & Read, 1999) and also are involved in inherited cancer syndromes (Varmus & Weinberg, 1993). For instance, in familial cases of retinoblastoma, one parent passes on a defective copy of a tumor-suppressor gene to the offspring. The offspring will be heterozygous for the defective allele, meaning that he or she will have one normal copy and one mutated copy of the gene. The single copy of a normal gene is powerful enough to maintain its growth-suppressing potential, and the cell retains healthy function. During development, however, a chance exists that the normal copy of the gene becomes defective from faulty cell division or other mechanisms. Defects such as these may occur spontaneously once in a million cell divisions (Varmus & Weinberg). Unrestricted cell proliferation occurs in the cell containing the two faulty versions of the tumor-suppressor gene, and, consequently, the cell becomes malignant.

Cancer: An Acquired or Hereditary Genetic Disease?

Cancer progression is a multistep genetic disease involving two or more independent events (DeVita et al., 2001). Development from normal tissue to invasive carcinoma takes place over 5–20 years and is a result of acquired somatic mutations or hereditary factors (DeVita et al.). Some cancers exist in both hereditary and sporadic forms (McKinnell, Parchment, Perantoni, & Pierce, 1999). In rare childhood cancers, 30% of tumors may arise in genetically predisposed individuals; however, hereditary factors may only be responsible for 5%–10% of common adult cancers (DeVita et al.). The genetic constitution of an individual can explain the discrepancy in cancer development. For example, 30% of women with a mutation in the BRCA1 gene will develop ovarian cancer, and women with common variants in the HRAS gene appear to have greater susceptibility to this malignancy (DeVita et al.).

Methods of Genetic Analysis

The detection of DNA mutations is important in understanding how a gene causes specific diseases (Jorde et al., 1999). Some of the basic techniques used in studying disease include enzymatic DNA digestion followed

ucts inhibit events leading toward cancer; therefore, they play an important role in the development of cancer as oncogenes (Strachan & Read; Varmus & Weinberg, 1993). If a tumor-suppressor gene is mutated, the gene becomes inactivated. The cell loses its ability to control cell growth, and its excessive proliferation thereby leads to

cancer. To use an analogy similar to the one before, tumor suppressor genes function like the brakes of a car. When mutated, the brakes fail and the car cannot slow down. In general, abnormal proliferation only occurs when two copies of the functionally recessive tumor-suppressor genes are inactivated, unlike oncogenes, where tumors develop as

by gel electrophoresis, DNA sequencing, or polymerase chain reaction. During gel electrophoresis, the digested DNA migrates through a gel, and fragment size can be determined. DNA sequencing is a technique that enables the exact nucleotide order of a gene to be determined. This could be compared to deciphering the words in a book by reading all the letters. Another exceedingly important method for the diagnosis of DNA mutation is polymerase chain reaction (PCR). PCR is used to amplify segments of DNA and is very useful in genetic screening, cancer diagnosis, and detection of residual disease in patients after cancer treatment (DeVita et al., 2001).

Fluorescent In Situ Hybridization

FISH is a molecular cytogenetic technique used to analyze genes and chromosomes and their aberrations occurring in cancer. The technique can identify specific chromosomal areas by attaching a probe to a targeted region of DNA. A probe is a cloned fragment of DNA of interest and is labeled with fluorescent color. The probe can identify a single gene or an entire chromosome (Lichter & Ried, 1994). The probe and target DNA are denatured, a process by which the double strand of DNA is unwrapped into two single strands. The denatured, single-stranded probe is placed onto the single-stranded target DNA. The probe anneals or attaches itself to the complementary area of DNA, thereby delineating the specific gene or chromosome (see Figure 3).

In a normal cell, the probe will hybridize in two places, reflecting the two homologous chromosomes (Jorde et al., 1999). If a probe only hybridizes to one chromosome, it indicates that a deletion of genetic material has occurred in the homologous chromosome. However, if an excess of chromosomal material exists, the probe will hybridize to that area in addition to two homologous chromosomes (Jorde et al.). For instance, when a probe is used to detect chromosome 8 in a normal cell, it highlights two chromosomes. Certain leukemias have a gain of chromosome 8 (Heim & Mitelman, 1995); therefore, the probe would illuminate three chromosomes 8 in an affected cell, clearly indicating a copy number increase. FISH is very useful in testing missing or additional chromosomal material, as well as chromosomal rearrangements (Jorde et al.).

Comparative Genomic Hybridization

CGH is a technique that allows one to screen entire tumor genomes to detect deletions or duplications of genetic material (Kallioniemi et al., 1992). This technique is especially beneficial because it allows one to detect aberrations in archived, previously fixed cancer samples.

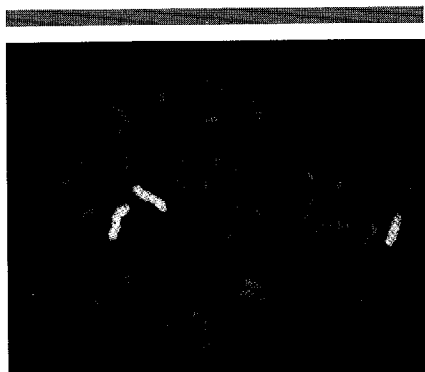
CGH requires the comparison of a tumor genome to a normal genome. The tumor DNA is labeled with green fluorescence, and the normal DNA is labeled with red fluorescence. A slide is prepared with normal target chromosomes. Based on the principles of FISH, the probe containing both the tumor and normal DNA are denatured into single strands and placed on the target slide containing denatured chromosomes. The single-stranded tumor and normal DNA "compete" to anneal together with the single strands of the target DNA on the slide. Consequently, if the chromosomes of both tumor and normal DNA are present in equal copy numbers, equal blends of the red and green colors are represented on the chromosomes. To use an analogy, if 10 children wearing green shirts (tumor DNA) and 10 children wearing red shirts (normal DNA) were playing a game of musical chairs, they would be distributed equally on the chairs (target chromosomes). If a segment or entire chromosome is deleted in the tumor genome, there would be an excess of the color red from the normal genome. So, if only 5 children with green shirts and 10 with red shirts remained, they would be unequally distributed and more red would be seen on the chairs. If, on the contrary, the tumor genome experiences a gain of DNA, such as an amplified oncogene, the color green would be greater in the region of amplifica-

tion. Thus, if 10 children with red shirts and 15 with green shirts remained, more children with green shirts than red would be sitting on the chairs. CGH, therefore, is an elegant method to identify and map cancer-causing chromosomal copy number changes (see Figure 4).

Spectral Karyotyping

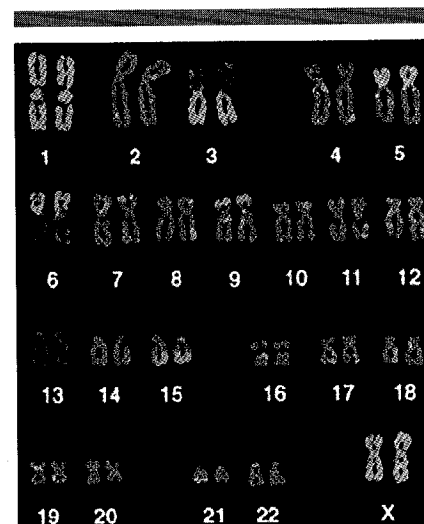
SKY is a novel technique allowing for the visualization of all human chromosomes in different colors (Schröck et al., 1996). Like CGH, this technique is used to screen the entire genome for chromosomal aberrations. In contrast to CGH, SKY also detects chromosomal aberrations, such as translocations, that do not affect the DNA copy numbers (see the CML Philadelphia chromosome in Figure 5).

SKY allows for each set of chromosomes to be "painted" in different colors. This is achieved by applying 24 chromosome-specific probes (Macville et al., 1997) so that chromosome 1 is yellow, chromosome 2 is red, and so on. Each set of chromosomes is discerned from the other according to color. SKY can be used to identify genetic aberrations, such as gains or losses of entire chromosomes and translocations. For example, in the case of a translocation, if a yellow chromosome 1 has a blue tip, it indicates that the translocated segment is coming from chromosome 9, which is blue. SKY can be remarkably helpful in examining complex



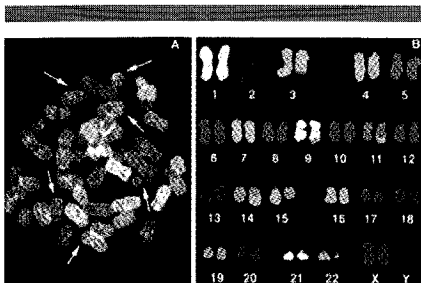
The hybridization pattern indicates three copies of this chromosome. Trisomies of chromosome 7 frequently are found in prostate and colorectal cancers.

FIGURE 3. FLUORESCENT IN SITU HYBRIDIZATION USING A PROBE SPECIFIC FOR CHROMOSOME 7



Blue indicates a balance between tumor and normal genomes, green reflects regions of DNA amplification in the tumor genome, and red shows the areas that are deleted in the tumor genome.

FIGURE 4. COMPARATIVE GENOMIC HYBRIDIZATION FLUORESCENCE RATIO IMAGE OF AN ADVANCED STAGE CERVICAL CARCINOMA



A, metaphase chromosomes. B, karyotypic analysis showing aberrations involving chromosomes 3, 11, 14, and 15, and the Philadelphia chromosome translocation from chromosome 22 to 9

FIGURE 5. SPECTRAL KARYOTYPING IN A CASE OF CHRONIC MYELOID LEUKEMIA

Note. From "Hidden Chromosomal Abnormalities in Hematological Malignancies Detected by Multicolor Spectral Karyotyping," by T. Veldman, C. Vignon, E. Schrock, J.D. Rowley, & T. Ried, *Nature Genetics*, 15, p. 406. Copyright 1997 by Nature Publishing Group. Reprinted with permission.

chromosomal arrangements with high accuracy, identifying marker chromosomes, and determining the chromosome of origin of each translocated segment (see Figure 5) (Veldman, Vignon, Schröck, Rowley, & Ried, 1997). SKY, therefore, facilitates the comprehensive evaluation of chromosomal aberrations in cancer.

Implications for Clinical Practice

The molecular cytogenetic techniques of FISH, CGH, and SKY are providing tremendous insights into genetic aberrations related to cancer. This is achieved specifically by illustrating complex translocations, small deletions, amplifications, and chromosomal copy number changes that can occur in a patient's cancer cells. These molecular cytogenetic techniques can help to translate the information derived from the Human Genome Project to the clinic. This will result in the better diagnosis of both solid tumors and hematologic malignancies.

Cancer is a multistep process and a disease of the DNA. To illustrate this point further, specific genetic aberrations occur during each tumor stage (Ried, Heselmeyer-Haddad, Blegen, Schröck, & Auer, 1999). Genetic instability increases significantly as the cells transform from premalignant lesions to invasive carcinomas. For instance, increases in copy number on chromosomes 7 and 20 occur in colorectal adenomas. These chromosomes contain oncogenes relevant for the development of colorectal cancer. After the transition to invasive carcinomas, however, additional genetic gains

occur on chromosome 13, again in loci of oncogenes, and losses on chromosome 8 in regions of tumor-suppressor genes.

Through the use of FISH, CGH, and SKY, molecular diagnostic tests can be developed using chromosomal markers targeting the genetic changes that are characteristic of each stage of the tumor. This will improve the diagnosis and staging of tumors, particularly in small, premalignant lesions that often are difficult to assess. As a result of early cancer detection, prognosis will improve and disease-free survival time will increase. Furthermore, an early diagnosis will reduce medical costs. The costs associated with cancer detection and treatment vary; however, estimates indicate that the medical cost for a patient with an early (stage I) breast cancer is \$33,419 compared to \$89,000 for a patient diagnosed at a locally advanced stage (Brown et al., 1999).

Recently, the possibilities and disadvantages of genetic testing for hereditary forms of cancer have caused much excitement. However, molecular cytogenetic techniques make it possible to test for nonhereditary tumors as well. For example, if the c-MYC gene (found on chromosome 8) is amplified in a suspicious breast lesion, a conclusion can be drawn that this lesion is neoplastic because c-MYC amplification is involved in tumor development.

The Role of the Oncology Nurse

Advances in molecular biology and genetics have provided an understanding of the cellular basis of cancer (Jorde et al., 1999). This understanding of cancer is providing a foundation of knowledge that is critical for oncology nurses. The blending of genetics

- Provide information about cancer risk factors, such as heredity, lifestyle, diet, hormones, environment, and occupation.
- Include information about cancer risk reduction, wellness promotion, and preventive measures.
- Discuss cancer surveillance methods, including self-examination information and recommended frequency of screening tests implicated for individuals of each age group and sex.
- Provide information about cancer counseling programs, including genetic counseling and support groups.
- Explain the genetic tests and research protocols when relevant.

FIGURE 6. PATIENT-EDUCATION POINTERS

Alliance of Genetics Support Groups

<http://www.geneticalliance.org>

American Cancer Society

www.cancer.org

Atlas of Genetics and Cytogenetics in Oncology and Hematology

<http://www.infobiogen.fr/services/chromcancer/>

Breast Cancer and Genetic Screening

<http://www.lbl.gov/Education/ELSI/screening-main.html>

Cancer Network

<http://www.cancernetwork.com/>

Genetics Education Center

<http://www.kumc.edu/gec/>

Genetics Societies Home Page

<http://www.faseb.org/genetics/>

Human Genome Project

<http://www.nhgri.nih.gov>

International Society of Nurses in Genetics

<http://nursing.creighton.edu/isong>

National Cancer Institute

<http://www.nci.nih.gov>

National Institutes of Health Glossary of Genetic Terms

http://www.nhgri.nih.gov/DIR/VIP/Glossary/pub_glossary.cgi

Oncolink: Genetics and Cancer

<http://www.oncolink.upenn.edu/causeprevent/genetics/>

Oncology Nursing Society

<http://www.ons.org>

FIGURE 7. INTERNET RESOURCES

with clinical management consequently will impact oncology nursing practice.

Nurses are an important part of the patient-education process because they can interpret and translate information based on genetic research to patients. Oncology nurses are compelled to help patients, families, and the public understand this information and how it relates to their own lives (Greco, 2000). Teaching should emphasize cancer prevention, wellness promotion, and screening for indicators of high risk (see Figure 6) (Gates & Fink, 1997).

The challenge exists, however, in keeping ahead of the growing body of information. Patients are aware of genetic information provided by the Internet and public information networks. Therefore, nurses must assess patients' and families' knowledge and assist in the correct interpretation of genetic information. Oncology nurses must provide psychosocial support, as well as resources and referrals related to hereditary cancers, to patients and families undergoing cancer screening and risk assessment (see Figure 7). By increasing their awareness in genetics and molecular testing and its impact on disease, oncology nurses

will be empowered to educate their patients, be advocates, and provide comprehensive care.

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For more information on this topic, visit the following Web sites:

[Atlas of Genetics and Cytogenetics in Oncology and Haematology](http://www.infobiogen.fr/services/chromcancer/)
<http://www.infobiogen.fr/services/chromcancer/>

[American Journal of Human Genetics](http://www.journals.uchicago.edu/AJHG/)
<http://www.journals.uchicago.edu/AJHG/>

[University of Washington: Cytogenetics Gallery](http://www.pathology.washington.edu/Cytogallery/)
<http://www.pathology.washington.edu/Cytogallery/>

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Rapid Recap

Molecular Cytogenetics and Gene Analysis: Implications for Oncology Nurses

- As members of the healthcare team, nurses must be knowledgeable about the rapidly emerging genetic information.
- Fluorescent in situ hybridization (FISH) is very useful in testing missing or additional chromosomal material, as well as chromosomal rearrangements.
- Comparative genomic hybridization (CGH) is a technique used to screen entire genomes to detect deletions or duplications of genetic material.
- Spectral karyotyping (SKY) can be helpful in examining complex chromosomal arrangements with high accuracy and in identifying marker chromosomes and chromosomal translocations.
- Through the use of FISH, CGH, and SKY, molecular diagnostic tests can be developed using chromosomal markers targeting the genetic changes that are characteristic of each stage of the tumor. This will improve the diagnosis and staging of tumors, particularly in small, premalignant lesions that often are ambiguous to assess.
- Oncology nurses are an important part of the patient-education process because they can interpret and translate information based on genetic research to patients and help patients, families, and the public to understand this information and how it relates to their own lives.